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Leukaemia Section
Short Communication

t(9;15)(p13;q24) PAX5/GOLGA6A

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Abstract

Review on t(9;15)(p13;q24) PAX5/GOLGA6A, with data on clinics, and the genes implicated.

Clinics and pathology

Disease

B-cell acute lymphoblastic leukemia (B-ALL)

Note

Was not taken into account in this review a case of diffuse large B-cell lymphoma in a 51 year-old male patient (Morrison et al., 1994).

Epidemiology

One case to date, a 21-year-old male patient with a CD10+ (B-II, common) ALL (Coyaud et al., 2010).

Prognosis

No data.

Cytogenetics

Cytogenetics morphological

There were additional numerial anomalies (+5, +21).

Genes involved and proteins

PAX5

Location

9p13.2

Protein

391 amino acids; from N-term to C-term, PAX5 contains: a paired domain (aa: 16-142); an octapeptide (aa: 179-186); a partial homeodomain (aa: 228-254); a transactivation domain (aa: 304-359); and an inhibitory domain (aa: 359-391). Lineage-specific transcription factor; recognizes the concensus recognition sequence GNCCANTGAAGCGTGAC, where N is any nucleotide. Involved in B-cell differentiation. Entry of common lymphoid progenitors into the B cell lineage depends on E2A, EBF1, and PAX5; activates B-cell specific genes and repress genes involved in other lineage commitments.

Activates the surface cell receptor CD19 and repress FLT3.

Pax5 physically interacts with the RAG1/RAG2 complex, and removes the inhibitory signal of the lysine-9-methylated histone H3, and induces V-to-DJ rearrangements.

Genes repressed by PAX5 expression in early B cells are restored in their function in mature B cells and plasma cells, and PAX5 repressed (Fuxa et al., 2004; Johnson et al., 2004; Zhang et al., 2006; Cobaleda et al., 2007; Medvedovic et al., 2011).

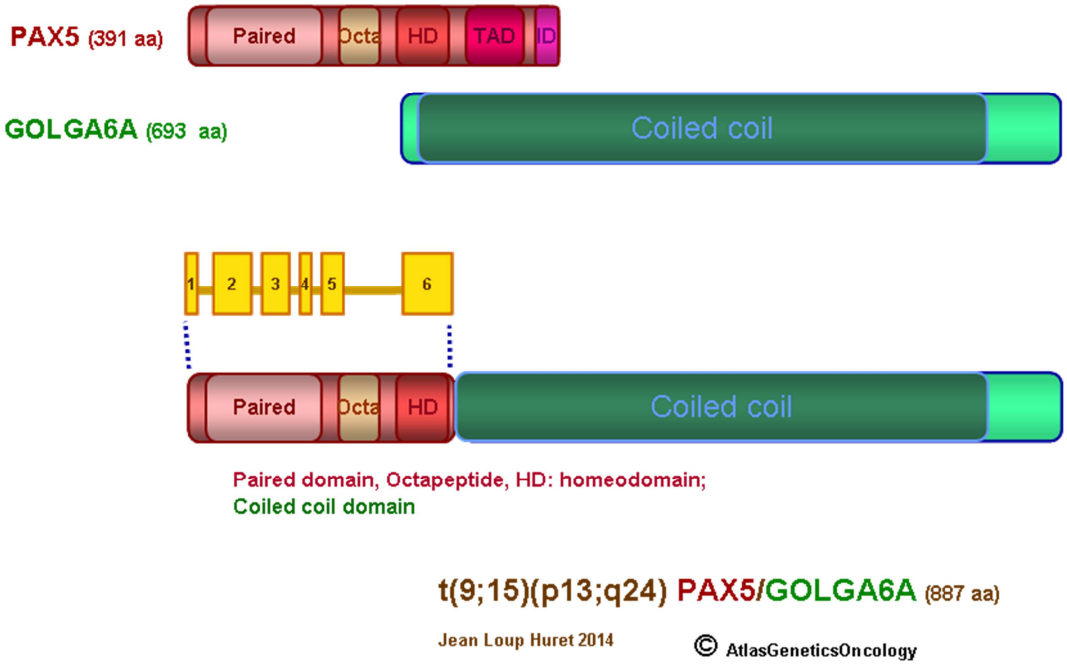
GOLGA6A

Location

15q24.1

Protein

693 amino acids (aa); contains a coiled coil domain (aa 14-611), with 11 leucine zipper motifs (aa 360-430).



PAX5/GOLGA6A fusion protein.

Member of the golgin family of proteins, localizes to the Golgi apparatus. Its function is poorly known (Gilles et al., 2000).

Result of the chromosomal anomaly

Hybrid gene

Description

Fusion of PAX5 exon 6 to GOLGA6A exon 3.

Fusion protein

Description

887 amino acids. The predicted fusion protein contains the DNA binding paired domain, the octapeptide, and the homeodomain of PAX5 and part of the coiled coil domain of GOLGA6A.

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